KAYSER-FLEISCHER RING: A VISUAL SIGN OF WILSON'S DISEASE.

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Abstract

Wilson's disease (WD) is a rare, treatable, autosomal recessive disease which leads to pathological copper accumulation in various organs and tissues[1]. The habitual clinical manifestations include hepatic, neurological, or psychiatric symptoms, often appearing early in life[2]. A common ophthalmological finding in this disease is the Kayser-Fleischer ring, resulting from copper deposition in Descemet's membrane of the cornea[3], as illustrated in the presented clinical case.

Keywords: Wilson's disease, Kayser-Feischer ring.

Introduction

Wilson's disease (WD) is an autosomal recessive inherited disorder, rare in the general population. It is characterised by deficient biliary excretion of copper, leading to its accumulation in tissues, mainly in the liver and central nervous system, with chronic liver disease, progressive neurological disorder and psychiatric illness, with onset in early life being characteristic[1]. Another typical manifestation is ophthalmological, with the characteristic Kayser-Fleischer ring, secondary to copper deposition in Descemet's membrane of the cornea[3]. We present a case of the debut of an advanced chronic liver disease secondary to Wilson's disease with this peculiarity.

Clinical case

A 22-year-old woman from Morocco, with no personal history of interest, presented to the emergency department with one week of abdominal distension and oedema of the lower limbs, weight loss, asthenia and hyporexia. Physical examination revealed oedema of the lower limbs, moderate ascites and jaundice, with a normal neurological examination. Laboratory tests showed total bilirubin levels of 15 mg/dL, elevated transaminases (AST/ALT <2), platelets and coagulopathy. Abdominal ultrasound showed a small, heterogeneous and

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Plaza Fernández A, Navarro Moreno E, Sánchez García O. Kayser-Fleischer ring: a visual sign of Wilson's disease. RAPD 2024;47(4)166-167. DOI: 10.37352/2024474.6

hyperechogenic liver, splenomegaly and multicompartmental ascites. Upper gastrointestinal endoscopy was performed with findings of small oesophageal varices and mild gastropathy of portal hypertension. The aetiological study was completed with negative serology and autoimmunity tests, with only low ceruloplasmin, so 24 hours urine copper test was requested. In the meantime, she was assessed by ophthalmology and the characteristic Kayser-Fleisher (KF) ring was observed (Figures 1 and 2). Finally, a cupruria of >40 mcg was obtained and the diagnosis of WD was established. Treatment with copper chelators was prescribed, with adequate tolerance, and the pre-transplant study was initiated.

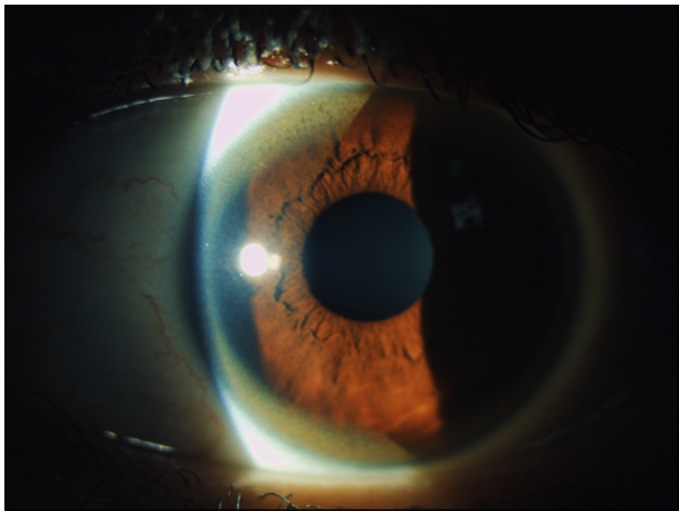

Figure 1. Kayser-Fleischer ring visible at the corneal limbus.

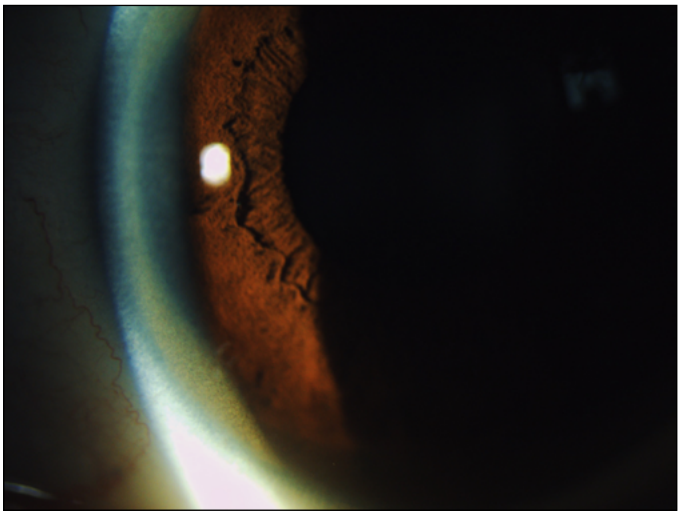

Figure 2. Kayser-Fleischer ring visible at the corneal limbus.

Discussion

Kayser-Fleischer ring is a characteristic ophthalmological finding, although not pathognomonic of WD, resulting from copper deposition in Descemet's membrane of the cornea[1]. It is present in almost all patients with WD and neurological involvement, but only in 50% with liver involvement. It also occurs in other pathologies such as primary biliary cholangitis or neonatal cholestasis[3].

Slit-lamp examination is essential for the diagnosis of Kayser-Fleischer ring, especially in the early stages[2]. They are usually asymptomatic and bilateral in onset, do not cause visual disturbances and resolve with medical treatment, 80% of them respond to chelator therapy within 3-5 years. However, they may recur with disease progression, functioning as valuable indicators of the patient's response to therapy and adherence to treatment[3].

WD is a rare but treatable inherited disorder, the early identification of which is crucial to prevent serious complications. This case illustrates the importance of a thorough diagnosis, including ophthalmological evaluation of the Kayser-Fleischer ring, a significant but not pathognomonic finding of the disease. Proper management of WD, with the use of copper chelators and continuous follow-up, can significantly improve the patient's quality of life and reduce associated morbidity and mortality. It is essential to maintain a high index of clinical suspicion and to perform a thorough evaluation in patients with unexplained liver symptoms to ensure early diagnosis and timely treatment.

Bibliography

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DOI: 10.37352/2024474.6